GENETICS

- Scientific study of heredity
- HUMAN GENETICS -Scientific study of variations and heredity in human beings
- MEDICAL GENETICS -Application of knowledge of human genetics for the practice of medicine and medical research

BRANCHES OF GENETICS

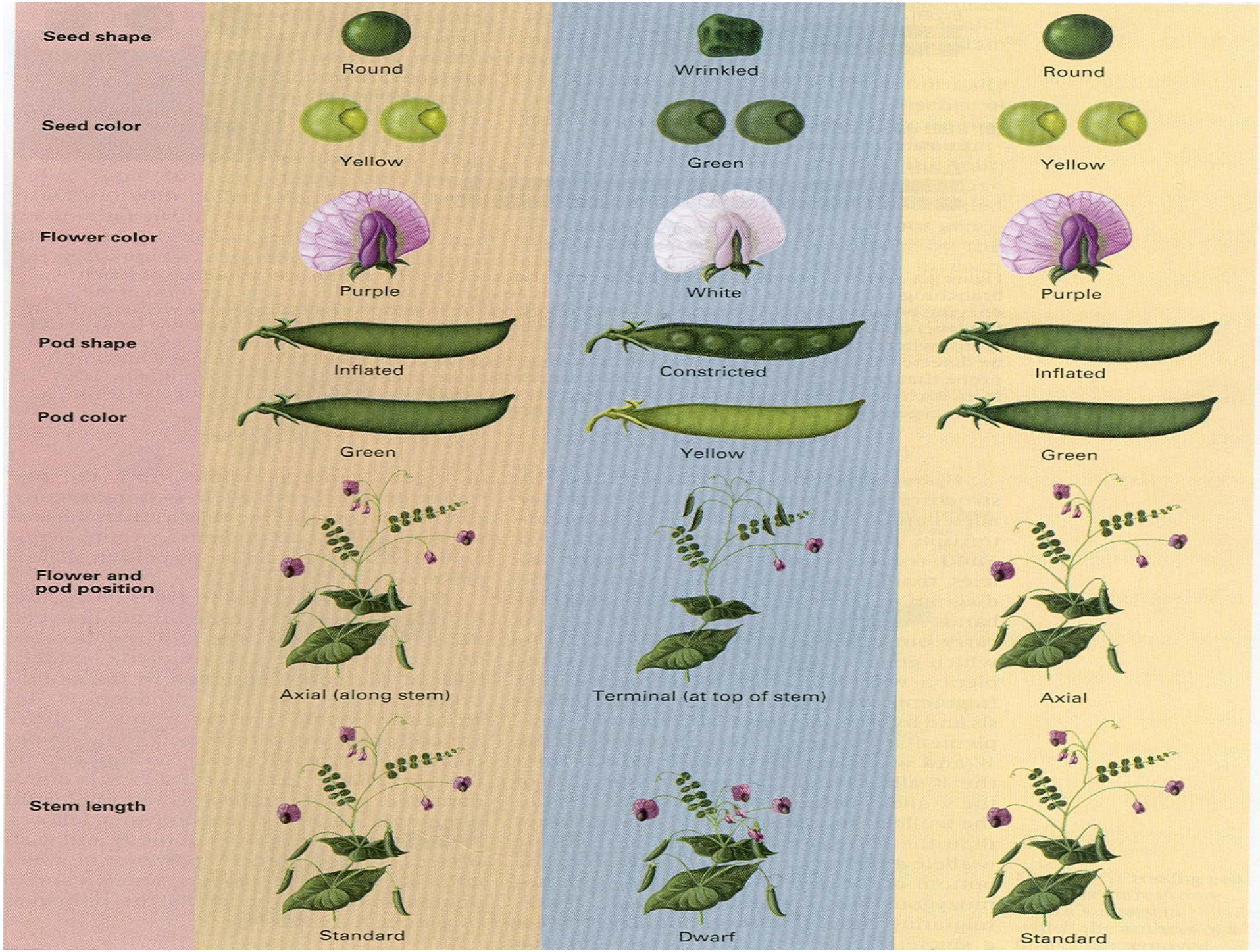
- CYTOGENETICS: Study of chromosomes
- MOLECULAR & BIOCHEMICAL GENETICS: Study of structure & function of genes
- PAPULATION GENETICS: Study of genetic variations in human population and factors that determine allele frequency
- DEVELOPMENTAL GENETICS – study of genetic control of development
- CLINICAL GENETICS – Diagnosis of genetic disease and care of patient with such disease.

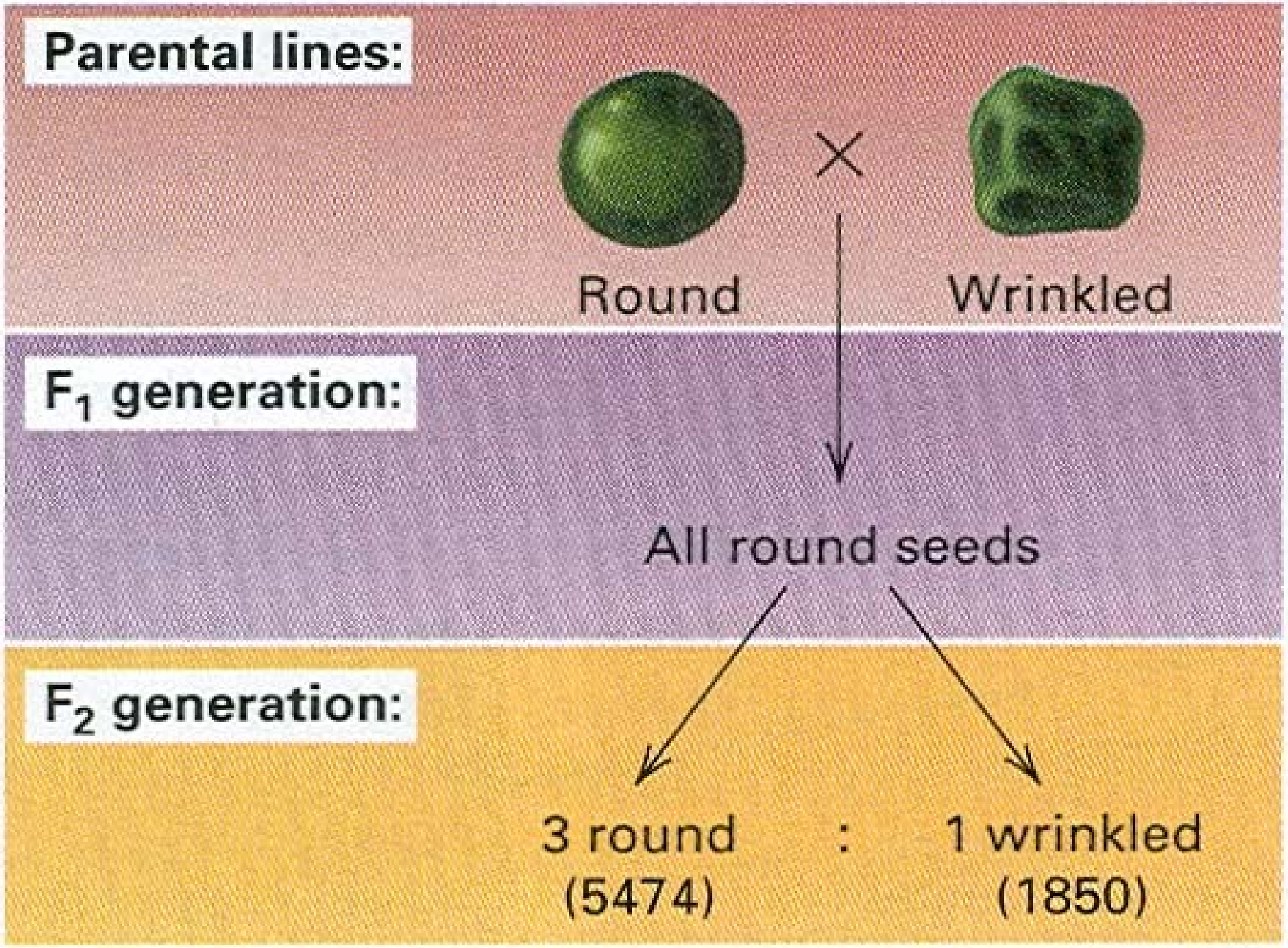
HISTORICAL ASPECTS

- Concept of heredity is around 6000 old.
- Haemophilia -First genetic disorder discovered 1500 years ago.
- Aristotle believed semen had ability to give life to embryo
- In 17th century Ragnier de Graff coined concept of fertilization.
- Equal contribution of parents to their offsprings was discovered by Maupertuis-a French naturalist

MENDEL'S LAWS OF INHERITANCE

- Gregor Mendel(1853) discovered principals of hereditary.
- He worked on few characters of sweet pea.

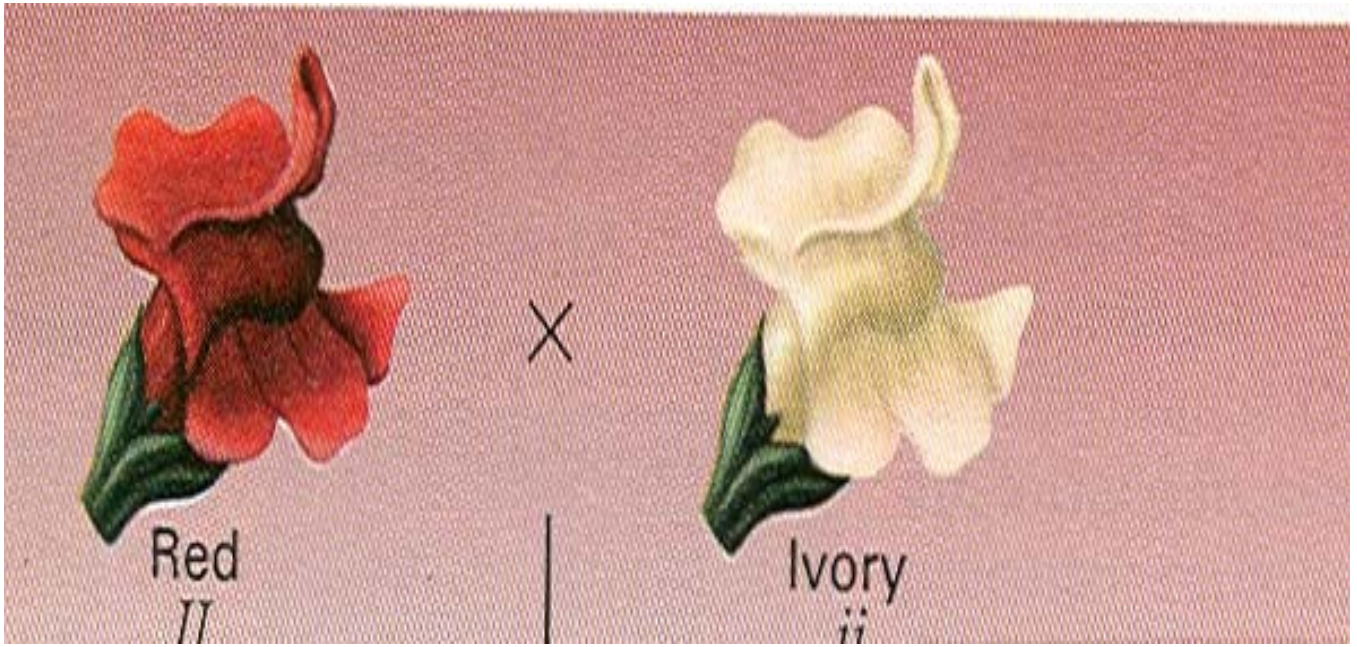

Seed shape
Round
Wrinkled
Round
Seed color
Yellow
Green
Yellow
Flower color
Purple
White
Purple
Pod shape
Inflated
Constricted
Inflated
Pod color
Green
Yellow
Green
Flower and pod position
Axial (along stem)
Terminal (at top of stem)
Axial
Stem length
Standard
Dwarf
Standard


Parental lines:
Round
×
Wrinkled
F1 generation:
All round seeds
F2 generation:
3 round
(5474)
:
1 wrinkled
(1850)

- LAW OF UNIT INHERITANCE: Inherited characteristics determined by pairs of hereditary elements
- LAW OF SEGREGATION: Two members of single pair of genes pass to different gametes during reproduction
- LAW OF INDEPENDENT ASSORTMENT: Members of different gene pairs assort independently of one another during gametogenesis

GENETIC TERMINOLOGY

- Genes – particulate hereditary element
- Alleles – alternative forms of a given gene
- Heterozygous – when members of a pair of allele are different
- Homozygous – when members of a pair of alleles are same
- Genotype – genetic constitution of an organism
- Phenotype – Observable property of an organism
- Dominance – expression of phenotype in heterozygous



CHROMOSOMAL BASIS OF INHERITANCE

- In 1860’s nucleus of cell was identified & considered responsible for heredity.
- Walther Flamming(1877) identified chromosomes in nucleus
- Sutton and Boveri(1903) found behavior of chromosomes during gametogenesis paralleled with Mendel’s heredity unit

CHEMICAL BASIS OF INHERITANCE

Avery, McLeod and McArthy worked on pneumococcus and discovered nucleic acid

- The nucleic acid forms structure of DNA
- DNA is the genetic material responsible for inheritance

CLINICAL APPLICATIONS

- Early diagnosis
- Better medical and nursing care can improve quality of life
- Genetic counseling and screening test
- Reproductive option for couple